IN THE CLAIMS

Claims 1-8 (cancel)

9 (withdrawn) A method for the diagnosis of diseases related to immune maturation and regulation of immune response towards self and nonself, characterized by detecting in a biological specimen the presence of a DNA sequence comprising the sequence id. no. 1 or a functional fragment or variant thereof, or a functionally equivalent isolated DNA-sequence hybridizable thereto.

10. (Withdrawn) A method according to claim 9, characterized in that the DNA sequence is associated with APECED.

11. (Withdrawn) A method according to claim 9 , characterized in that the DNA sequence includes a gene defect responsible for APECED.

12. (Withdrawn) A method according to claim 11, characterized in that the gene defect to be detected includes a"C"to"T" transition resulting in the"Arg"to "Stop"nonsense mutation at amino acid position 257 and/or a"A"to"G" transversion resulting in the"Lys"to"Glu"missense mutation at amino acid position 42.

13. (Withdrawn) A method according to claim 9, characterized in that DNA techniques are used for the detection.

14. (Withdrawn) A method according to claim 9, characterized in that the detection takes advantage of TaqI or another enzyme cleaving at recognition site 5'-TCGA-3'digestion.

15. (Withdrawn) A method according to claim 9, characterized in that the disease is autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

16. (Withdrawn) A method for the diagnosis of diseases related to immune maturation and regulation of immune response towards self and nonself, characterized by detecting in a biological specimen the presence or the absence of a protein comprising the sequence id. no. 1, or a functional fragment thereof having the sequence according to sequence id. no. 4, or a functional fragment thereof having the sequence according to sequence id. no. 6.

17. (Withdrawn) A method according to claim 16, characterized in that the protein is associated with APECED.

18. (Withdrawn) A method according to claim 16 , characterized in that the disease is autoimmune polyendocrinopathy-candidiasisectodermal dystrophy (APECED).

19. (Withdrawn-Currently amended) The use of the DNA sequence according to claim 1 claim 25 in the diagnosis of diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

20. (Withdrawn-Currently amended) The use of the protein according to claim 28 ~~claim 5~~ in the diagnosis of diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

21. (Withdrawn-Currently amended) The use of the DNA sequence according to ~~claim 1~~ claim 25 for the preparation of a medicament useful in a gene therapy method of diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathycandidiasis-ectodermal dystrophy (APECED).

22. (Withdrawn-Currently amended) The use of the DNA sequence according to ~~claim 1~~ claim 25 in the treatment of diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

23. (Currently amended) A reagent ~~Reagents~~ capable of reacting with a ~~the~~ DNA sequence characterized by comprising ~~the sequence id. no. 1 or a functional fragment~~ SEQ ID NO:1 or variant thereof ~~encoding a protein having the same functional activity~~, which variant includes a mutation responsible for autoimmune polyendocrinopathy-candidiasis ectodermal dystrophy (APECED), said mutation resulting in R257X mutation in the protein sequence encoded by SEQ ID NO:1.

24. (Withdrawn) Reagents according to claim 23, characterized in that they are antibodies.

25 (new) An isolated nucleic acid molecule comprising the sequence of SEQ ID NO:1 or a variant thereof, which variant includes a mutation responsible for autoimmune polyendocrinopathy-candidiasis ectodermal dystrophy (APECED), said mutation resulting in R257X mutation in the protein sequence encoded by SEQ ID NO:1.

26 (new) An isolated nucleic acid molecule comprising the sequence of SEQ ID NO:1 or a variant thereof, which variant includes includes a mutation responsible for autoimmune polyendocrinopathy-candidiasis ectodermal dystrophy (APECED), said mutation resulting in K42E mutation in the protein sequence encoded by SEQ ID NO:1. or an functionally equivalent isolated DNA sequence hybridizable thereto or the protein of claim 5 or with reagents therewith.

27 (new) An isolated nucleic acid molecule comprising the sequence of SEQ ID NO:1.

28 (new) A protein comprising an amino acid sequence of SEQ ID NO:2 .

29 (new) A protein comprising an amino acid sequence of SEQ ID NO:2 or a variant thereof, which variant includes a R257X mutation responsible for

autoimmune polyendocrinopathy-candidiasis ectodermal dystrophy (APECED).

30 (new) A protein comprising an amino acid sequence of SEQ ID NO:2 or a variant thereof, which variant includes a K42E mutation responsible for autoimmune polyendocrinopathy-candidiasis ectodermal dystrophy (APECED).

31 (new). A protein comprising an amino acid sequence of SEQ ID NO:2 or a variant thereof of SEQ ID NO:4.

32 (new). A protein comprising an amino acid sequence of SEQ ID NO:2 or a variant thereof of SEQ ID NO:6.

33 (new) A protein according to claim 29 comprising a structural motif selected from the group consisting of the PHD finger motif (PHD), the LXXLL motif (L), proline-rich region (PRR) and cystein-rich region (CRR).

34 (new) A protein according to claim 30 comprising a structural motif selected from the group consisting of the PHD finger motif (PHD), the LXXLL motif (L), proline-rich region (PRR) and cystein-rich region (CRR).

35 (new) A reagent capable of reacting with a DNA sequence characterized by comprising SEQ ID NO: 1 or a variant thereof, said variant includes a mutation responsible for autoimmune polyendocrinopathy-candidiasis ectodermal dystrophy (APECED), said mutation resulting in K42E mutation in the protein sequence encoded by SEQ ID NO:1 .